

A case report - goldenhar's syndrome

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Abstract

A boy aged 2 years 6 months was reported to our district hospital with a rare syndrome of goldenhar having multiple symptoms. He had facial asymmetry, scoliosis, eye problems like limbaldermoid, hypoplastic mandible with bilateral preauricular skin tag and there is history of delayed dentition and the baby was operated during neonatal period for tracheoesophagialfistulla. Child also had absent Right Kidney.

Keywords: Goldenhar syndrome, Facial asymmetry, Preauricular tag.

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INTRODUCTION

Goldenhar syndrome is a rare hereditary condition due to multifactorial etiopathology leading to disturbance of blastogenesis¹. Goldenhar first described this condition in 1952 as a combination of several anomalies such as , dermal epibulbar tumours. Preauricular appendices, facial asymmetry. Gorlin in 1963 also described the Goldenhar syndrome in association with heart disease, hypoplasia the zygomatic, mandibular and maxillary bones², eye anomalies like coloboma of upper eyelid, iris, choroidea and retina, ear anomalies like microtia, anotia, preauricular skin tags, blind fistulas and other external ear manifestations^{3, 4, 5}. The incidence of this condition varies from 1 in 3500 to 1 in 5600 live births³. The Male: Female ratio is approximately 3:2. Cases may report with malformations of cardiac, renal, vertebral, ear, central nervous system and others skeletal anomalies⁶. A few cases of Orofacial anomalies have also been described.

We report a case of a with Goldenhar syndrome with various clinical manifestations as described above.

CASE REPORT

A 2 years 6 months old boy was brought to outpatient department with a history of cough and cold, on examination child looks comfortable, afebrile, runny nose, respiratory rate of 20 cycles / min, pulse rate of 106/min and BP 90/60 mm of Hg in right upper limb. Facial asymmetry with right side hypoplasia with limbaldermoid (ophthalmologist opinion) which was not disturbing his vision, Bilateral preauricular skin tag without any sinuses with no hearing disturbance. There was a right sided delayed eruption of mandibular teeth and there was a gross scoliosis towards the left side which was also evident by X-rays spine. There was no cardiac abnormalities, respiratory sounds were normal and child was neurologically normal but ultrasonography of the abdomen shows absent right kidney. The child was operated for tracheoesophageal fistula during neonatal period and the child's speech and hearing was intact. The parents were nonconsanguinously married without any family history of similar complaints and this is the only child. No maternal history of any medications and exposure to radiations.

Investigations

Blood routine was normal and renal functions were within normal limits. 2D Echo was normal, X-ray of face showed right mandibular hypoplasia. Ultrasound of abdomen showed absent right kidney. Dentist opinion was taken and advised for correction of maloccluded teeth.



DISCUSSION

The patient presented to us with various clinical manifestations in the form of occulofacioauricular and vertebral anomalies as described above as in Goldenhar syndrome⁸. Facial asymmetry and hypoplasia of the mandible are typical features of Goldenhar syndrome⁹. Which were present in our case. On the other hand the presence of epibulbar dermoid tumour is variable² and it is usually associated with bilateral preauricular skin tags which is observed in our case. There were no hearing abnormalities and no facial nerve palsy¹⁰ in this syndrome cardiac abnormality was observed in 5 to 58 percent of patients¹¹ which was not found in our case. Renal anomalies were common in this syndrome¹² and in our case there was absent right kidney with normal renal functions. There was no family history and this case could be a sporadic. Drugs like primidone, thalidomide, vitamin -A, cocaine exposure in pregnancy and also tobacco and herbicides have been associated with this syndrome¹³.

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